

# Hirschsprung's Disease Causing Constipation in an Infant

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## CASE REPORT

A 12-month-old boy with past medical history significant for constipation and gastroesophageal reflux presented to our emergency room with symptoms of persistent vomiting progressing into hematemesis over a 2-day period. Physical examination was benign. Hemoglobin level was reassuring at 11.6 gm/dL. Patient was admitted for further evaluation and management. An upper gastrointestinal study with oral contrast revealed normal anatomy. As he was advanced to a regular diet, he started having emesis again 24 hours after the upper gastrointestinal study was completed. His abdominal exam at that point was significant for severe distention. Therefore, an abdominal x-ray was obtained that revealed a large amount of retained barium contrast within moderately dilated descending and severely dilated proximal sigmoid colon with a normal caliber rectum (Figure 1). A surgical consultation was placed, and the patient underwent a full-thickness rectal biopsy that revealed absence of ganglion cells and abnormal acetylcholinesterase staining confirming the diagnosis of Hirschsprung's disease (HD).

Hirschsprung's disease is a congenital disorder that is characterized by the complete absence of ganglion cells in myenteric and submucosal plexus for a variable length of colon.<sup>1</sup> Ganglion cells act as points within the enteric nervous system to help coordinate and facilitate bowel relaxation. With their absence, the aganglionic areas of the bowel become spastic, thus causing distal intestinal obstruction.<sup>2</sup> A transition zone is the term applied to the region where a marked change in caliber occurs, with the dilated normal colon above, and the narrowed aganglionic colon below forms. Prompt diagnosis is key in order to avoid significant complications such as toxic megacolon or acute enteritis.<sup>3</sup> Obtaining a good HPI is critical when HD is suspected. A barium enema done beyond first year of age, demonstrating that a transition zone is a hallmark for HD. Although this is a highly reliable sign, failure to visualize a transition zone is common with barium enemas and thus does not rule out the presence of the disease. Anorectal manometry is another option that is becoming a much more prominent and popular method for diagnosis. However, anorectal biopsy continues to be the gold standard.<sup>4</sup> In conclusion, the use of an upper gastrointestinal



**Figure 1.** Abdominal x-ray obtained 24 hours after upper gastrointestinal study was performed. X-ray reveals a large amount of retained contrast material within moderately dilated descending and moderate to severely dilated proximal sigmoid colon. Concern for HD.

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series followed by an abdominal x-ray the following day is an interesting option since it eliminates risk of deflating the colon during the barium enema and missing a transition zone.

## DISCLOSURES

Author contributions: Both authors contributed equally to the manuscript and serve as article guarantors.

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Informed consent was obtained for this case report.

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